

## Celidocranial Dysostosis Followed for 25 Years. Presentation of A Case

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## 1. Abstract

Cleidocranial Dysostosis is an autosomal dominant scale dysplasia. We present a case of Cleidocranial Dysostosis of a female patient with deformities in the development of the osteomyoarticular system due to clavicular agenesis, deficiency in mandibular development and dentition, which was diagnosed very early in childhood. Follow-up is carried out in the orthopedic consultation for 25 years, reaching the main conclusion that the development of this entity affects little the psychosocial development of the patients.

## 2. Introduction

Cleidocranial Dysostosis (CCD) is an autosomal dominant scale dysplasia of skeletal and dental tissues, characterized by aplasia or hypoplasia of clavicles, multiple Wormian bones, failure of mesenchymal ossification, delayed dental eruption, supernumerary teeth and other skeletal abnormalities. Little amount of information has been published, in particular a comprehensive study to describe skeletal abnormalities. CCD is a congenital disorder with

an incidence of 1 in 1,000,000 newborns and more than the 1,000 cases reported so far [1,2].

This condition was described for the first time in 1766 for Morand and later in 1897 by Pierre Marie and Sinton, in 1898 it was called CCD, a rare skeletal alteration that particularly damages the bones of the head and the clavicular area, it is also known under the name of Marie Sinton Disease, Scheuthauer Marie Sinton Disease and / or Cleidocranial Dysostosis Syndrome [3, 4].

This article presents a patient with this rare familial pathology, little addressed in the literature.

## 3. Case Presentation

Information of the patient. 27-year-old white female patient, born from a eutocic delivery at 41 weeks, with a family history: a healthy father and a mother with a history of presenting difficulties in the osteomyoarticular system.

The patient was seen for the first time at 18 months of age, when she went to the orthopedic consultation brought in by her par-

ents for presenting visible alterations in the body, especially at the shoulder level, when examining her, the absence of clavicular bone was found, disorder in the closure of fontanelles and delayed teething. She underwent X-rays, observing the presence of underdeveloped Wormian bones, supernumerary teeth, abnormalities of dental eruption and absent clavicles (Figure 1).



**Figure 1:** Polydontia and delayed dentition, absence of clavicular bones. X-ray performed in 1994 at 18 months of age.

Diagnosis of CCD or Marie Sainton disease is made. The patient is closely followed by the orthopedic consultation for 25 years, where psychomotor development was assessed, not showing any disorder in her intellectual development and in the development of the musculoskeletal system, little development of stature was found, brachycephaly, discrete exophthalmia, wide neck and reduction of the lateral diameter of the upper portion of the thorax, in addition to the absence of the clavicular bones.

### 3.1. Diagnostic Evaluation

Bone defects in the patient mainly involve the clavicles, head and jaw, the muscles associated with the clavicles are abnormal with little development, long neck, absence of the clavicles with unusual mobility of the shoulders, the patient can bring the shoulders closer together in front of the chest, this is an important characteristic of Marie Sainton disease, it is considered pathognomonic for the diagnosis. Although there are defects and variations of the associated muscles, the function is relatively normal, the rib cage is small and bell-shaped, the ribs are short, (Figure 2) the legs are abnormally small, and also defects in the hands and feet, abnormally long second metacarpal.



**Figure 2:** Chest X-ray. Bell-shaped chest, low-set shoulders, and no clavicles. (X-ray performed in 2020 at the age of 27)

<http://www.acmcasereport.com/>

### 3.2. Therapeutic Intervention

The therapy was expectant with the development of the patient, as well as her orientation in the rehabilitation and adaptation of her to the physical conditions of her person.

### 3.3. Monitoring and Results

At this time, the patient is 27 years old and has developed her life normally, without significant limitations for her personal and social development.

### 4. Discussion

CCD is clinically characterized by presenting clavicle hypoplasia or aplasia, with decreased mobility of the shoulders, imperfection and / or delay of the ossification of the fontanelles that generates the voluminous configuration of the head or the depression of the line of the front area, as well as pointed jaw, hypertelorism and hyperdontia which fills the maxillary area and dental malalignment in the oral cavity [5]. In the case presented, these characteristics can be appreciated.

The diagnosis, as in the case presented, is basically clinical and radiological, which provides proof of pathognomonic identity of the disease caused by clavicle agenesis, hyperdontia, and delayed closure of the fontanelles [6].

Cleidocranialdysostosis is a genetic defect in the CBFA1 gene, also called RUMX2, once 6p21 was located on the chromosome, it acts by mutating chondrocytes and osteoblast non-differentiation [7, 8].

It is known that mutations in the fibroblastic receptor apparatus of growth factor 1,2 and 3 (FGFR1, 2 and 3), and that mutations in the transcription factor RUNX2 (CBFA1) cause cleidocranialdysostosis. It is a disorder that involves a mutation in the transcription factor, which controls the differentiation of osteoblast precursor cells, which is essential for membranous ossification, as well as endochondral ossification [9, 10]. In the case studied chromosomal studies were not possible.

In Cuba there are several information about families with this disease, Arocha Rodríguez and collaborators present 15 cases in a family in Havana, [11] Márquez Ibáñez N and his team have studied a family for eight generations and show the data of 17 people in Holguín province, [3] also MarchenaIdavoy JL, shows a case of absence of clavicle as the fundamental symptom for diagnosis, in most of these cases [4]

Genetic counseling for the family about the disease is fundamental and the therapeutic management is based on it, it causes a 50% risk of affecting a child of each patient, we clarify that this disease varies in its clinical presentation and that its main complications are in the young child's stage of development and should be assessed early.

### 5. Conclusions

CCD is an intrafamilial disorder of clinical variability, several

bones are affected in these patients, the most common condition is the absence of the clavicular bone, there is also involvement of the upper jaw and dentition disorders. It must be managed by a multidisciplinary team of specialists made up of an orthopedic, radiologist, geneticist and pediatricians, who evaluate patients to provide the best care. It does not affect the psychosocial development of the patients.

**6. The contribution of the Authors:** The authors carried out attention to the case, writing of the work and the analysis of the bibliography.

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